

Benjamin J O'callaghan

List of Publications by Year in descending order

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Version: 2024-02-01

6
papers

159
citations

1937685

4
h-index

1872680

6
g-index

7
all docs

7
docs citations

7
times ranked

286
citing authors

#	ARTICLE	IF	CITATIONS
1	<i>ANGPTL6</i> Genetic Variants Are an Underlying Cause of Familial Intracranial Aneurysms. <i>Neurology</i> , 2021, 96, e947-e955.	1.1	6
2	Brownâ€™Violettoâ€™Van Laere and Fazioâ€™Londe syndromes: <i>SLC52A3</i> mutations with puzzling phenotypes and inheritance. <i>European Journal of Neurology</i> , 2021, 28, 945-954.	3.3	5
3	The startle disease mutation Î±1S270T predicts shortening of glycinergic synaptic currents. <i>Journal of Physiology</i> , 2020, 598, 3417-3438.	2.9	2
4	Bi-allelic JAM2 Variants Lead to Early-Onset Recessive Primary Familial Brain Calcification. <i>American Journal of Human Genetics</i> , 2020, 106, 412-421.	6.2	47
5	An update on the genetics, clinical presentation, and pathomechanisms of human riboflavin transporter deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 598-607.	3.6	65
6	A novel <i>ATP1A2</i> mutation in a patient with hypokalaemic periodic paralysis and CNS symptoms. <i>Brain</i> , 2018, 141, 3308-3318.	7.6	26